More than 1 in 200 people have an inherited cardiovascular condition

CLINICAL IMPLICATIONS OF CARDIOLOGY GENETIC TESTING

The underlying molecular etiology of heritable arrhythmias, cardiomyopathies, aortopathies, and other heart conditions can be identified through genetic testing. Genetic testing is an important aid in the diagnosis of these diseases and provides important prognostic and therapeutic information.[1-7]

DIAGNOSIS

- Confirms a clinical diagnosis
- Differentiates from other causes
- Fulfills diagnostic criteria for complex heart disorders
- Identifies at-risk asymptomatic and pre-symptomatic family members

PROGNOSIS

- Identifies patients and family members at highest risk for life-threatening arrhythmias, aortic dissections, or heart failure
- Predicts response to pharmacotherapies

MANAGEMENT

- Informs avoidance of certain medications and guides lifestyle modifications to avoid triggering cardiac events
- Aids in the decision to place implantable cardiac defibrillator or pacemaker and directs treatment options such as enzyme replacement therapy, early surgical intervention, and heart transplantation
- Enables appropriate screening intervals and interventions for at-risk family members

GUIDELINES AND CONSENSUS STATEMENTS RECOMMEND GENETIC TESTING

Guidelines and consensus statements endorsed by numerous professional societies, including ACCF, AHA, HFSA, HRS, and PACES, recommend cardiology genetic testing.[4-7]

Heart Rhythm Society (2011) Consensus Recommendations[4]

	CONDITION	PATIENTS WITH ESTABLISHED OR SUSPECTED DIAGNOSIS	FAMILY
ARRHYTHMIAS	Brugada syndrome	**Can be useful**	**Recommended**
	Catecholaminergic polymorphic ventricular tachycardia	**Recommended**	**Recommended**
	Long QT syndrome	**Recommended:** For diagnosed/suspected patients or asymptomatic patients with idiopathic, serial QTc values >480 ms (prepuberty) or >500 ms (adults) **May be considered:** For asymptomatic patients with idiopathic, serial QTc values >460 ms (prepuberty) or >480 ms (adults)	**Recommended**
	Short QT syndrome	**May be considered**	**Recommended**
	Postmortem SIDS/SUDS	**May be considered:** In the setting of autopsy-negative sudden unexplained death syndrome (SUDS)	**Recommended**
CARDIOMYOPATHIES	Arrhythmogenic right ventricular cardiomyopathy	**Can be useful:** For patients satisfying 2010 task force diagnostic criteria **May be considered:** For patients with 1 major or 2 minor criteria **Not recommended:** For patients with only a single, minor criterion	**Recommended**
	Hypertrophic cardiomyopathy	**Recommended**	**Recommended**
	Dilated cardiomyopathy	**Can be useful**	**Recommended**
	Left ventricular noncompaction	**Can be useful**	**Recommended**

Abbreviations: ACCF, American College of Cardiology Foundation; AHA, American Heart Association; HFSA, Heart Failure Society of America; HRS, Heart Rhythm Society; PACES, Pediatric & Congenital Electrophysiology Society

INVITAE CARDIOLOGY GENETIC TESTS

ARRHYTHMIA AND CARDIOMYOPATHY

Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel	up to 148 genes

ARRHYTHMIA

Invitae Arrhythmia Comprehensive Panel	up to 73 genes
Invitae Arrhythmogenic Cardiomyopathy Panel	up to 24 genes
Invitae Brugada Syndrome Panel	up to 20 genes
Invitae Catecholaminergic Polymorphic Ventricular Tachycardia Panel	8 genes
Invitae Long QT Syndrome Panel	up to 17 genes
Invitae Short QT Syndrome Panel	up to 6 genes

CARDIOMYOPATHY

Invitae Cardiomyopathy Comprehensive Panel	up to 105 genes
Invitae Arrhythmogenic Cardiomyopathy Panel	up to 23 genes
Invitae Dilated Cardiomyopathy Panel	up to 69 genes
Invitae Hypertrophic Cardiomyopathy Panel	up to 60 genes
Invitae Left Ventricular Noncompaction Panel	up to 19 genes
Invitae Transthyretin Amyloidosis Test	1 gene
Invitae Hereditary Hemochromatosis Panel	5 genes
Invitae RASopathies Comprehensive Panel	18 genes

CARDIOMYOPATHY AND SKELETAL MUSCLE DISEASE

Invitae Cardiomyopathy and Skeletal Muscle Disease Panel	up to 158 genes

AORTOPATHY AND CONNECTIVE TISSUE DISORDERS

Invitae Aortopathy Comprehensive Panel	up to 25 genes
Invitae Ehlers-Danlos Syndrome Panel	14 genes
Invitae Loeys-Dietz Syndrome Panel	up to 6 genes
Invitae Marfan Syndrome Test	1 gene

FAMILIAL HYPERCHOLESTEROLEMIA

Invitae Familial Hypercholesterolemia Panel	4 genes

PULMONARY HYPERTENSION

Invitae Pulmonary Arterial Hypertension Panel	up to 9 genes
Invitae Hereditary Hemorrhagic Telangiectasia Panel	up to 5 genes
Invitae Capillary Malformation-Arteriovenous Malformation Syndrome Test	up to 4 genes

CONGENITAL HEART DISEASE

Invitae Congenital Heart Disease Panel	42 genes
Invitae CHARGE Syndrome Test	1 gene
Invitae Costello Syndrome Test	1 gene
Invitae Holt-Oram Syndrome Test	1 gene
Invitae RASopathies Comprehensive Panel	18 genes
Invitae Sotos Syndrome Test	1 gene

REFERENCES

1. Marcus FI, *et al. Circulation*. 2010;121(13):1533-41.
2. MacCarrick G, *et al. Genet Med*. 2014;16(8):576-87.
3. Loeys BL, *et al. Journal of Medical Genetics*. 2010;47(7):476-85.
4. Ackerman MJ, *et al. Heart Rhythm*. 2011;8(8):1308-39.
5. Priori SG, *et al. Heart Rhythm*. 2013;10(12):1932-63.
6. Gersh BJ, *et al. Circulation*. 2011;124(24):2761-96.
7. Hershberger RE, *et al. J Card Fail*. 2009;15(2):83-97.

INVITAE

HEADQUARTERS & LABORATORY | 1400 16th Street, San Francisco, CA 94103 | www.invitae.com | **IN THE US** | clientservices@invitae.com | p: 800-436-3037 | f: 415-276-4164
OUTSIDE THE US | globalsupport@invitae.com or visit www.invitae.com/contact |

P/N F139-11